Connective Tissue Disorders: Deletion/Duplication Panel

**Test Code:** MD270  
**Turnaround time:** 2 weeks  
**CPT Codes:** 81228 x1

### Condition Description

Connective tissues provide a framework and structural support for the body, protect organs, connect body tissues, and store energy. Connective tissue disorders are a heterogeneous group of more than 200 conditions that often involve the joints, muscles, and skin. Heritable connective tissue disorders include both soft tissue disorders, characterized by excessive skin laxity, joint hypermobility, and easy bruising, as well as skeletal dysplasias that affect bone development.

Diagnosis of heritable connective tissue disorders may be challenging due to extensive clinical variability, phenotypic overlap, or atypical presentation. However, making a specific diagnosis is important given that some of these disorders feature life-threatening complications, such as aortic root rupture, that require careful medical surveillance and monitoring. In addition, treatment options are available for some connective tissue disease.

**Reference:**  
Bateman et al. (2009), Nat Rev Genet. 10:173-183.

### Genes

**ACTA2, ACVR1, ADAMTS2, ATP6V0A2, CBS, CHST14, COL11A1, COL1A1, COL1A2, COL2A1, COL3A1, COL5A1, COL5A2, ELN, FBLN5, FBN1, FBN2, FKBP14, MYLK, NOTCH1, PKD2, PLOD1, PRDM5, SLCA2A10, SLCA39A13, SMAD3, TGFBR1, TGFBR2, ZNF469**

### Indications

This test is indicated for:

- Confirmation of a clinical diagnosis of connective tissue disorder.

### Methodology

**Deletion/Duplication Analysis:** DNA isolated from peripheral blood is hybridized to a CGH array to detect deletions and duplications. The targeted CGH array has overlapping probes which cover the entire genomic region. Please note that a "backbone" of probes across the entire genome are included on the array for analytical and quality control purposes. Rarely, off-target copy number variants causative of disease may be identified that may or may not be related to the patient's phenotype. Only known pathogenic off-target copy number variants will be reported. Off-target copy number variants of unknown clinical significance will not be reported.

**Detection**

**Deletion/Duplication Analysis:** Detection is limited to duplications and deletions. The CGH array will not detect point or intronic mutations. Results of molecular analysis must be interpreted in the context of the patient's clinical and/or biochemical phenotype.

### Specimen Requirements

Submit only 1 of the following specimen types

**Type: Whole Blood**

Specimen Requirements:

In EDTA (purple top) tube:  
Infants (2 years): 3-5 ml  
Older Children & Adults: 5-10 ml.

Specimen Collection and Shipping: Ship sample at room temperature with overnight delivery.

**Type: Isolated DNA**

Specimen Requirements:
In microtainer: 10 ug

Isolation using the Qiagen™ Puregene kit for DNA extraction is recommended.

Specimen Collection and Shipping: Refrigerate until time of shipment in 100 ng/µl of TE buffer. Ship sample at room temperature with overnight delivery.

**Related Tests**

- Connective Tissue Disorders: Sequencing Panel